

**Table of Disorders Screened by the Program**

Condition	Incidence	Symptoms if not Detected	Treatment
<b>Other Disorders</b>			
<p><b>Biotinidase Deficiency:</b></p> <p>A condition in which the body is unable to use biotin, a B vitamin.</p>	<p>1 in 60,000 births</p> <p>(1 in 106,773 births in Hawaii)</p>	<p>Mental retardation, seizures, skin rash, loss of hair, death</p>	<p>Supplement with biotin</p>
<p><b>Congenital Adrenal Hyperplasia (CAH):</b></p> <p>A condition in which the adrenal glands are unable to produce normal amounts of certain hormones.</p>	<p>1 in 13,700 births</p> <p>(1 in 21,354 births in Hawaii)</p>	<p>Salt wasting, dehydration, shock in infants</p> <p>Abnormal genital organs in females</p>	<p>Glucocorticoid and/or mineralcorticoid</p>
<p><b>Congenital Hypothyroidism:</b></p> <p>A condition in which the thyroid gland cannot make enough thyroid hormone for normal body and brain growth.</p>	<p>1 in 4,485 births</p> <p>(1 in 2,512 births in Hawaii)</p>	<p>Mental retardation, other brain damage, growth delay</p>	<p>Thyroid hormone replacement</p>
<p><b>Cystic Fibrosis:</b></p> <p>A condition that causes thick, sticky mucus and fluids to build up in certain organs in the body, especially the lungs and the pancreas.</p>	<p>1 in 3,000 births</p> <p>(1 in 32,147 births in Hawaii)</p>	<p>Pancreatic insufficiency and lung disease</p>	<p>Enzyme replacement and organ transplant</p>
<p><b>Galactosemia:</b></p> <p>A condition in which the body cannot break down a sugar (galactose) found in milk.</p>	<p>1 in 60,000 births</p> <p>(1 in 106,773 births in Hawaii)</p>	<p>Severe brain damage, kidney damage and eye abnormalities in neonates, death</p>	<p>Strict galactose-free diet</p>
<p><b>Hemoglobinopathies (including Sickle Cell):</b></p> <p>Conditions in which</p>	<p>Sickle cell disease:</p> <p>1 in 15, 000</p>	<p>Sickle cell disease:</p> <p>Anemia, painful crises, death</p>	<p>Sickle cell disease:</p> <p>Penicillin</p>

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abnormal hemoglobin in red blood cells may cause anemia	births  (1 in 35,591 births in Hawaii)		
<b>Amino Acid Disorders</b>			
<b>Arginase Deficiency:</b>  A condition in which the body cannot get rid of a toxic substance called ammonia	Rare  (No cases in Hawaii)	Developmental delay, seizures, hyperactivity, ataxia	Restrict arginine and protein in diet  Supplement with amino acids other than arginine  Sodium benzoate therapy
<b>Argininosuccinate Lyase Deficiency (ASA):</b>  A condition in which the body cannot get rid of a toxic substance called ammonia	1 in 70,000  (No cases in Hawaii)	Mental retardation, potential lethal coma, seizures, anorexia, vomiting, lethargy	Restrict protein in diet  Supplement with arginine
<b>Citrullinemia:</b>  A condition in which the body cannot get rid of a toxic substance called ammonia	n/a  (No cases in Hawaii)	Mental retardation, potential lethal coma, seizures, anorexia, vomiting, lethargy	Low protein diet  Sodium benzoate, phenylacetate, arginine
<b>Homocystinuria:</b>  A condition in which the body cannot break down several amino acids in protein foods	1 in 200,000 births  (No cases in Hawaii)	Heart disease, stroke, possible mental retardation, psychiatric problems	Low methionine diet  Supplement with pyridoxine, L-cysteine, and betaine
<b>Phenylketonuria (PKU):</b>  A condition in which the body cannot break down one of the amino acids found in protein foods	1 in 15,900 births  (1 in 71,182 births in Hawaii)	Severe mental retardation, seizures	Low phenylalanine diet
<b>Tyrosinemia Types I and II:</b>  A condition in which the	1 in 100,000 births  (1 in 1,846	Liver disease, kidney problems, seizures, rickets	Low phenylalanine and tyrosine diet  Liver transplant if

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body cannot break down several amino acids in protein foods	French Canadian births		necessary
<b>Organic Acid Disorders</b>			
<p><b>Beta-Ketothiolase Deficiency:</b></p> <p>A condition in which the body cannot break down and get rid of certain organic acids</p>	<p>Rare</p> <p>(No cases in Hawaii)</p>	<p>Recurrent, severe metabolic acidosis</p>	<p>Sodium bicarbonate, IV fluids</p> <p>Possible dialysis</p> <p>Supplement with carnitine</p>
<p><b>Glutaric Acidemia Type I:</b></p> <p>A condition in which the body cannot break down and get rid of certain organic acids</p>	<p>1 in 30,000 livebirths</p> <p>(More common in people of Amish ancestry)</p>	<p>Neurological deterioration, muscle weakness, seizures, possible dystonic cerebral palsy</p> <p>Some people may have no symptoms</p>	<p>Restrict lysine and tryptophan in diet</p> <p>Supplement with riboflavin and carnitine</p>
<p><b>Isobutyryl CoA Dehydrogenase Deficiency:</b></p> <p>A condition in which the body cannot break down and get rid of certain organic acids</p>	<p>Very rare</p> <p>(No cases in Hawaii)</p>	<p>Heart problems</p>	<p>Carnitine supplementation</p>
<p><b>Isovaleric Acidemia:</b></p> <p>A condition in which the body cannot break down and get rid of certain organic acids</p>	<p>1 in 50,000 births</p> <p>(1 in 53,431 births in Hawaii)</p>	<p>Vomiting, lack of appetite, lethargy, neuromuscular irritability, hypothermia</p>	<p>Protein-restrictive diet</p> <p>Supplement with carnitine and glycine</p>
<p><b>Malonic Aciduria:</b></p> <p>A condition in which the body cannot break down and get rid of certain organic acids</p>	<p>Rare</p> <p>(No cases in Hawaii)</p>	<p>Developmental delay, vomiting, seizures, cardiomyopathy, hypoglycemia</p>	<p>Avoid fasting</p> <p>Restrict fats in diet</p>
<p><b>Maple Syrup Urine Disease (MSUD):</b></p>	<p>1 in 150,000 births</p>	<p>Neonatal coma, convulsions, mental retardation, death</p>	<p>Diet low in branched chain amino acids</p>

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A condition in which the body cannot break down several amino acids in protein foods.	(1 in 53,486 births in Hawaii)		
<b>Methylmalonic Acidemias:</b>  A condition in which the body cannot break down and get rid of certain organic acids	1 in 50,000 to 1 in 100,000 births  (No cases in Hawaii)	Lethargy, vomiting, dehydration, respiratory distress, muscle weakness, coma, seizures, developmental delay	Low-protein diet and/or restriction of isoleucine, valine, and threonine
<b>Multiple Carboxylase Deficiency:</b>  A condition in which the body cannot break down and get rid of certain organic acids	1 in 87,000 births  (1 in 106,463 births in Hawaii)	Seizures, immune system impairment, skin rashes, hair loss, hearing loss, mental retardation	Biotin supplementation
<b>Propionic Acidemia:</b>  A condition in which the body cannot break down dietary fats to make energy	1 in 35,000 to 1 in 75,000 births  (No cases in Hawaii)	Mental retardation, seizures, movement disorders, coma, sudden death	Avoid fasting, low protein diet  Supplement with cornstarch, carnitine, and biotin  Antibiotic and human growth hormone treatment
<b>2-Methyl-3-Hydroxybutyryl CoA Dehydrogenase Deficiency:</b>  A condition in which the body cannot break down and get rid of certain organic acids	Rare  (No cases in Hawaii)	Developmental delay	In progress
<b>2-Methylbutyryl CoA Dehydrogenase Deficiency:</b>  A condition in which the body cannot break down and get rid of certain	Rare  (No cases in Hawaii)	Lethargy, irritability, coma	Dietary restrictions

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organic acids			
<b>3-Hydroxy-3-Methylglutaryl (HMG) CoA Lyase Deficiency:</b>  A condition in which the body cannot break down dietary fats to make energy	Rare  (No cases in Hawaii)	Persistent vomiting, muscle weakness, lethargy, seizures, coma	Avoid fasting, low fat, low protein, high carbohydrate diet  Supplement with carnitine and glucose
<b>3-Methylcrotonyl CoA Carboxylase Deficiency (3MCC):</b>  A condition in which the body cannot break down and get rid of certain organic acids	Rare  (No cases in Hawaii)	Muscle weakness and atrophy, seizures, dermatological changes	Dietary restrictions  Supplement with carnitine and/or biotin
<b>3-Methylglutaconyl CoA Hydratase Deficiency:</b>  A condition in which the body cannot break down and get rid of certain organic acids	Rare  (No cases in Hawaii)	Delayed motor development, short attention span, delayed development of speech	Still in development
<b>Fatty Acid Oxidation Disorders</b>			
<b>Carnitine Uptake/Transport Defects:</b>  A condition in which the body cannot break down dietary fats to make energy	Rare  (1 in 17,743 births in Hawaii)	Developmental delay, muscle weakness  Possible coma and death	Avoid fasting, low fat diet  Supplement with carnitine
<b>Glutaric Acidemia, Type II:</b>  A condition in which the body cannot break down dietary fats to make energy.	Rare  (No cases in Hawaii)	Muscle weakness, nausea, vomiting  Possible seizures, coma, and death	Avoid fasting, low fat diet  Supplement with carnitine
<b>Long Chain 3-hydroxyacyl-CoA Dehydrogenase</b>	Rare  (More common in	Developmental delay, muscle weakness, possible liver failure	Avoid fasting  Supplement with carnitine,

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<p><b>Deficiency (LCHAD):</b></p> <p>A condition in which the body cannot break down dietary fats to make energy.</p>	<p>those with Finnish ancestry)</p>		<p>cornstarch, MCT, and DHA</p>
<p><b>Medium Chain acyl-CoA Dehydrogenase Deficiency (MCAD):</b></p> <p>A condition in which the body cannot break down dietary fats to make energy.</p>	<p>1 in 15,000 births  (More common in Northern Europeans)</p>	<p>Development delay, seizures, coma, sudden death</p>	<p>Avoid fasting, low fat diet  Supplement with carnitine and cornstarch</p>
<p><b>Short Chain acyl-CoA Dehydrogenase Deficiency (SCAD):</b></p> <p>A condition in which the body cannot break down dietary fats to make energy.</p>	<p>Rare  (1 in 106,643 births in Hawaii)</p>	<p>Developmental delay, muscle weakness  Can have no symptoms or problems</p>	<p>Diet low in fats  Supplement with carnitine</p>
<p><b>Very Long Chain acyl-CoA Dehydrogenase Deficiency (VLCAD):</b></p> <p>A condition in which the body cannot break down dietary fats to make energy.</p>	<p>Rare  (1 in 35,487 births in Hawaii)</p>	<p>Heart problems, liver problems, sudden infant death</p>	<p>Avoid fasting, avoid certain fatty foods  Supplement with cornstarch, MCT, and possibly carnitine  IV glucose during illness</p>